# prothrombin deficiency

Prothrombin deficiency is a bleeding disorder that slows the blood clotting process. People with this condition often experience prolonged bleeding following an injury, surgery, or having a tooth pulled. In severe cases of prothrombin deficiency, heavy bleeding occurs after minor trauma or even in the absence of injury (spontaneous bleeding). Women with prothrombin deficiency can have prolonged and sometimes abnormally heavy menstrual bleeding. Serious complications can result from bleeding into the joints, muscles, brain, or other internal organs. Milder forms of prothrombin deficiency do not involve spontaneous bleeding, and the condition may only become apparent following surgery or a serious injury.

## Frequency

Prothrombin deficiency is very rare; it is estimated to affect 1 in 2 million people in the general population.

## Genetic Changes

Mutations in the *F2* gene cause prothrombin deficiency. The *F2* gene provides instructions for making the prothrombin protein (also called coagulation factor II), which plays a critical role in the formation of blood clots in response to injury. Prothrombin is the precursor to thrombin, a protein that initiates a series of chemical reactions to form a blood clot. After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

*F2* gene mutations reduce the production of prothrombin in cells, which prevents clots from forming properly in response to injury. Problems with blood clotting can lead to excessive bleeding. Some mutations drastically reduce the activity of prothrombin and can lead to severe bleeding episodes. Other *F2* gene mutations allow for a moderate amount of prothrombin activity, typically resulting in mild bleeding episodes.

## Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Other Names for This Condition

- dysprothrombinemia
- factor II deficiency
- hypoprothrombinemia

## Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Prothrombin deficiency, congenital
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0020640/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Factor II deficiency
  https://medlineplus.gov/ency/article/000549.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

## Additional Information & Resources

MedlinePlus

- Encyclopedia: Factor II deficiency
  https://medlineplus.gov/ency/article/000549.htm
- Health Topic: Bleeding Disorders
  https://medlineplus.gov/bleedingdisorders.html

Educational Resources

- Disease InfoSearch: Factor II Deficiency
  http://www.diseaseinfosearch.org/Factor+II+Deficiency/2708
- Disease InfoSearch: Inherited hypoprothrombinemia
  http://www.diseaseinfosearch.org/Inherited+hypoprothrombinemia/3828
- MalaCards: prothrombin deficiency
  http://www.malacards.org/card/prothrombin_deficiency
- Merck Manual Home Edition for Patients and Caregivers: How Blood Clots
  http://www.merckmanuals.com/home/blood-disorders/blood-clotting-process/how-blood-clots
- Orphanet: Congenital factor II deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=325
- University of Iowa Health Care: Prothrombin Gene Mutation
  https://www.healthcare.uiowa.edu/labs/lentz/Information_For_Patients/PDF/Prothrombin%20Gene%20Mutation%20Brochure.pdf

Patient Support and Advocacy Resources

- Canadian Hemophilia Society
  http://www.hemophilia.ca/en/bleeding-disorders/other-factor-deficiencies/factor-ii-deficiency--prothrombin-deficiency/
- National Hemophilia Foundation: Factor II Deficiency
  https://www.hemophilia.org/Bleeding-Disorders/Types-of-Bleeding-Disorders/Other-Factor-Deficiencies/Factor-II

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22prothrombin+deficiency%22+OR+%22Hypoprothrombinemias%22+OR+%22factor+II+deficiency%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28prothrombin+deficiency%5BTIAB%5D%29+OR+%28factor+II+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- COAGULATION FACTOR II
  http://omim.org/entry/176930

## Sources for This Summary


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